

SPINAL MUSCULAR ATROPHY

TYPE 2 | TYPE 3 | TYPE 4

Spinal Muscular Atrophy (SMA) is a type of childhood motor neurone disease.

SMA has a continuum from the most severe (Type 1) to the mildest form (Type 4) and the individual types are defined by the maximum motor skills gained.

INCIDENCE: 1 : 10,000 live births

AVERAGE AGE OF DIAGNOSIS: SMA 2 - 9 to 18 months (can sit independent when placed)

SMA 3 - 2 to 7 years (learn to walk independently)

SMA 4 - 16 years to adult (normal motor milestones)

OCCURRENCE: males and females affected equally

LIFE EXPECTANCY: ranges from early childhood to adulthood depending on severity, respiratory, nutritional and orthopaedic complications

Spinal Muscular Atrophy (SMA) is a genetic condition caused by a faulty gene carried by each parent that is then passed on to the child. SMA is caused by a lack of a protein called survival motor neuron (SMN), in the motor neurons within the spinal cord that control voluntary muscle movement. Without the SMN protein to support growth of the motor neurons coming from within the spinal cord they do not survive, and this results in decreased muscle function as the muscle cells die (atrophy). There is no current cure.

SMA is a progressive condition that primarily affects the muscles for movement and breathing. Children with SMA can have periods of relative stability. Weakness becomes more noticeable with growth. Characteristics can include tremor of the fingers, breathing issues, feeding difficulties and skeletal abnormalities (such as scoliosis and hip dislocation). The muscles that control breathing and coughing are significantly weak and the risk to children and adults with SMA2 is complications from coughs, colds, chest infections and pneumonia which can quickly become life threatening. The brain is not affected in SMA and those affected are usually intelligent, engaging and intuitive.

Infants with SMA2 will usually sit unsupported and some may crawl but they do not progress to walking. SMA2 can be very subtle and delays in motor skills may not to be evident until around 12 – 18 months of age. Children with SMA3 will gain the ability to walk independently, however the period of walking may decline as they get older and they may even cease walking all together by the time they are adolescents. SMA4 becomes evident

in adulthood and those affected will have a normal life expectancy and maintain most of their motor skills well into old age.

Children and adults with SMA require services and equipment to ensure they can interact with their surroundings, maintain good nutritional status, optimise their breathing and support their positioning. In order to gain independence children with SMA2 will require mobility aides from as early as 18 months of age. Children with SMA3 may still require mobility aides at later stages. Adults with SMA4 rarely require mobility aides.

Planning ahead for services & equipment is essential, particularly given the progressive yet unpredictable nature of SMA across the spectrum

As people with SMA age their needs will change, and the amount of support and care will increase. It is important that they are assessed at regular intervals to plan their future medical, physical, social and psychological supports. Medical complications can progress; therefore, the following areas should be monitored: respiratory care; sleep studies; non-invasive mechanical respiratory support; speech, swallowing and eating; continence issues, feeding devices and equipment; spine deformities and related surgeries.

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Genetic testing and counselling should be offered to people with SMA, parents and family members for future pregnancy planning. Parents and families experience enormous grief and loss in the dealing with the diagnosis and change in function of their child with SMA2 and SMA3. The knowledge that their child may have a shortened life expectancy can cause parental anxiety and counselling may be helpful for families. The need for "end-of-life" care planning should be discussed when deemed appropriate and referral to palliative care services made. Enough support needs to be provided to the person with SMA so that their carer can have a break.

It is important that service provision is centered around quality of life outcomes & meets the needs of the individual

Families and individuals affected by chronic conditions need to be empowered in order to have control over their life choices. Through seeking guidance and support regarding care options that are available, individuals will be empowered to make the right decisions for themselves. Despite the physical challenges that children and adults with SMA face, there are many opportunities available to support those affected with SMA to have a productive and rewarding life.

CONSIDERATIONS FOR PLANNING SHOULD INCLUDE BUT ARE NOT LIMITED TO THE FOLLOWING:

- A consistent General Practitioner (GP)
- Specialists for monitoring and medical care planning eg. neurologist, respiratory physicians
- Support coordination for complex care needs
- Physiotherapy programs eg. hydrotherapy, stretching, cough assist, chest physio
- Occupational therapy to support daily living activities eg. to increase independence, equipment assessment
- Assistive technology eg. manual wheelchair, power wheelchair, shower chair, hoist, slings
- Orthotics eg. leg splints for night-time and ankle-foot orthoses (AFO's)
- Comfort/sleeping eg. pressure relieving mattress, electronic bed with high/low options
- Home modifications for access to all areas eg. ramps, bathrooms, kitchen, outdoors
- Speech therapy for swallowing and speech problems
- Nutritional support eg. dietician, feeding pumps & consumables, high calorie formulas
- Assistive ventilation devices eg. CPAP or BiPAP machines, cough assist machines
- Adapted sporting and recreation equipment eg. boccia equipment, sports wheelchair
- Vehicle modifications/transport eg. taxis to school, work and community
- Support workers eg. showering, toileting, meal preparation, feeding, accessing community
- Opportunities to socialise and be away from home eg. MDNSW Camps, accessible travel
- Surgery and hospital admission eg. scoliosis rods, permanent feeding tubes (PEGs)
- Professional psychological support eg. counsellor or clinical psychologist
- Future goal planning eg. independent living, study, work choices
- Financial entitlements eg. Centrelink, Medicare, Companion Card

USEFUL RESOURCES

MDNSW www.mdnsw.org.au

MDNSW NDIS Toolkit www.mdnsw.org.au/our-services/support-coordination-your-choice/ndis-faqs

The Loop - Your Neuromuscular Resource Hub www.theloopcommunity.org